

BD-STEPS II Birth Defects Case Definitions

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Explanation/Glossary

BIRTH DEFECT & DEFINITION

- The specific birth defect and its definition are listed.
- Other names for the defect are given as well as the defect group if applicable.

TYPES & DEFINITIONS

- The types of the specific birth defect (if they exist) and their definitions are listed.

INCLUSIONS

The standard inclusion criteria for the defect are listed below; any exceptions to the standard inclusions are listed for that specific defect(s).

The standard inclusions are as follows:

- In general, case infants must have one or more of the specific birth defects listed in the abstraction instructions.
- The etiology of the defect(s) must be unknown or uncertain.
- Cases can be live born or stillborn infants ≥ 20 weeks' gestation or with birth weight at least 500 grams, or prenatally diagnosed and terminated fetuses at any gestational age.
- Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect will only be accepted as cases for a small number of defects; this information is listed in each defect category under the inclusion/exclusion sections.
- Cardiac defects must be confirmed by echocardiography (echo), catheterization (cath), surgery or autopsy; include cases diagnosed by prenatal ultrasound/fetal echo only if done by a pediatric cardiologist or in a prenatal diagnosis center with expertise in this area.
- Fetuses/infants with "apparently balanced" chromosomal translocations and inversions are included in the study.

EXCLUSIONS

The standard exclusion criteria for the defect are listed; any exceptions to the standard exclusions are listed for that specific defect(s).

The standard exclusions are as follows:

- In general, fetuses/infants diagnosed with chromosomal/microdeletion/microduplication disorders or single gene disorders are excluded.
 - Cases with CHARGE phenotype will be assessed in the same way as those with other syndromes based on the results of mutation testing and clinical findings.
- Cardiac defects that are clinically-diagnosed, i.e., using only physical exam, chest radiography, ECG, are excluded.

- Autopsy cases in which maceration or disruption (from termination techniques) preclude definitive diagnosis are excluded; this does not apply to defects for which prenatal diagnosis is accepted under certain circumstances (see INCLUSIONS).
- Conjoined and acardiac twins are excluded.
- Fetuses/infants with a microdeletion or microduplication detected by a chromosome microarray (aCGH) are excluded from the study if the copy number variant (CNV) is known to be pathogenic for the subject's phenotype. Subjects with a CNV that is benign, not associated with the subject's phenotype, or of unknown significance will be included in the study.
 - Clinical significance of CNVs will be decided by the study geneticist reviewing cases at each site. In assessing significance, consideration will be given to the following factors:
 - Results of parental studies that assess presence/absence of the CNV, when available
 - Reports of association of the CNV with a clinical phenotype related to the subject's phenotype
 - Presence within the CNV of a gene that is known or strongly suspected to be dosage sensitive and related to the subject's phenotype
 - Size of the CNV – Note that while most clinically significant CNVs are larger than 400 kb, this is not always the case. No specific size criteria are designated for inclusion/exclusion.
 - When a CNV's clinical significance is not clear-cut, the geneticist will consult with two geneticists from other study sites to review the evidence and come to a mutual decision about inclusion/exclusion of the subject. Rationale for this decision will be documented in the comment field for the case in the clinical tool.

ICD-9-CM CODES (for case finding)

- The ICD-9-CM codes for the specific birth defect to be used for case finding are listed.
 - Cases identified through ICD-9-CM codes should be reviewed to determine if they truly have a birth defect eligible for the study.
- The ICD-9-CM codes are used for the discharge diagnoses and are generally less specific than a surveillance system's BPA or modified codes.

ICD-10-CM CODES (for case finding)

- The ICD-10-CM codes for the specific birth defect to be used for case finding are listed.
 - Cases identified through ICD-10-CM codes should be reviewed to determine if they truly have a birth defect eligible for the study.
- The ICD-10-CM codes are used for the discharge diagnoses and are generally less specific than a surveillance system's BPA or modified codes.

CBDRP CODES (for eligible defects)

The CBDRP codes for the eligible defect(s) are listed.

1. Spina Bifida

BIRTH DEFECT & DEFINITION

- SPINA BIFIDA--herniation of the meninges and/or spinal cord tissue through a bony defect of spine closure

TYPES & DEFINITIONS

- SPINA BIFIDA CYSTICA, SPINA BIFIDA aperta -- synonyms for spina bifida.
- MYELOSCHISIS, MYELOYDYSPLASIA, etc. -- synonyms for spina bifida.
- MENINGOMYELOCELE/MYELOMENINGOCELE -- herniation of meninges and spinal cord tissue; 90% of lesions are of this type.
- MENINGOCELE -- herniation of meninges without spinal cord tissue
- RACHISCHISIS -- spine defect without meninges covering the neural tissue
- LIPOMENINGOMYELOCELE/LIPOMENINGOCELE -- lipomatous (fatty) tissue associated with a bony defect of the spine and herniation of meninges or spinal cord tissue, usually closed and located in the lumbosacral region
- MYELOCYSTOCELE -- cystic lesion of the spinal cord central canal with herniation through a spinal defect
- OPEN LESION -- neural tissue open to the environment or covered by a membrane only; 90% of lesions are of this type.
- CLOSED LESION -- neural tissue covered by normal skin
- LEVEL OF LESION -- highest and lowest vertebrae: cervical (C), thoracic (T), lumbar (L), sacral (S), cervicothoracic (CT), thorcolumbar (TL), lumbosacral (LS)

INCLUSIONS

- All cases including those prenatally diagnosed that do not have a postnatal examination to confirm the defect

EXCLUSIONS

- Spina bifida occulta
- Primary tethered cord
- Syringomyelia (hydromyelia)
- Diastematomyelia
- Diplomyelia
- Caudal lipomatous lesions not documented to involve neural tissue
- Iniencephaly -- a rare neural tube defect involving the occiput and inion, resulting in extreme retroflexion of the head variably combined with an occipital encephalocele or rachischisis of the cervical and thoracic spine
- Craniorachischisis -- anencephaly with contiguous rachischisis
- Any type of spina bifida with coexisting anencephaly
- Arnold-Chiari malformation without spina bifida, with or without hydrocephalus

ICD-9-CM CODES (for case finding)

- SPINA BIFIDA WITH HYDROCEPHALUS -- 741.00-741.03
- SPINA BIFIDA WITHOUT MENTION OF HYDROCEPHALUS -- 741.90-741.93

ICD-10-CM CODES (for case finding)

- SPINA BIFIDA -- Q05.0-Q05.9
- ARNOLD-CHIARI SYNDROME WITH SPINA BIFIDA -- Q07.01
- ARNOLD-CHIARI SYNDROME WITH SPINA BIFIDA AND HYDROCEPHALUS -- Q07.03

CBD RP CODES (for eligible defects)

- 741x0x: Meningomyelocele/myelomeningocele
- 741x1x: Meningocele
- 741x2x: Myelocele
- 741x3x: Myelocystocele
- 741x4x: Lipomeningomyelocele
- 741x5x: Lipomeningocele
- 741x6x: Rachischisis
- 741x8x: Other specified spina bifida
- 741x9x: Unspecified spina bifida
- 7410xx: Arnold Chiari malformation ± hydrocephalus, open lesion
- 7411xx: Arnold Chiari malformation ± hydrocephalus, closed lesion
- 7412xx: Arnold Chiari malformation ± hydrocephalus, unspecified open/closed lesion
- 7413xx: Hydrocephalus, other (aqueduct of Sylvius) or NOS, open lesion
- 7414xx: Hydrocephalus, other (aqueduct of Sylvius) or NOS, closed lesion
- 7415xx: Hydrocephalus, other (aqueduct of Sylvius) or NOS, unspecified open/closed lesion
- 7417xx: No mention hydrocephalus, open lesion
- 7418xx: No mention hydrocephalus, closed lesion
- 7419xx: No mention hydrocephalus, unspecified open/closed lesion
- 741xx1: Highest level, cervical
- 741xx2: Highest level, thoracic
- 741xx3: Highest level, lumbar
- 741xx4: Highest level, sacral
- 741xx9: Highest level, unspecified

2. Anophthalmia/microphthalmia

NOTE: These terms may be seen with the ending "ia", "os" or "us"

BIRTH DEFECT & DEFINITION

- ANOPHTHALMIA -- total absence of the eye tissue or apparent absence of the globe in an orbit that otherwise contains normal adnexal structures
- MICROPHTHALMIA -- reduction in the volume of the eye, usually characterized by corneal diameter less than 10 mm or anteroposterior globe diameter less than 20 mm

TYPES & DEFINITIONS

- TRUE OR PRIMARY ANOPHTHALMIA -- as above; occurs when there is complete failure of formation of the primary optic vesicle, usually bilateral; when unilateral, may have contralateral microphthalmia; verified only when histologic/microscopic exam shows that all ocular tissue is absent
- MICROPHTHALMIA -- categories: colobomatous (associated with uveal, iris, choroid and/or optic nerve colobomas) or noncolobomatous
- NANOPHTHALMIA -- microphthalmic eye with normal intraocular structures; this is a distinct genetic malformation

INCLUSIONS

- All cases must include diagnosis by an ophthalmologist or confirmation by prenatal or postnatal MRI, surgical pathology, autopsy

EXCLUSIONS

- Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect
- "Small eyes" or "small palpebral fissures" unless there is confirmation of anophthalmia or microphthalmia
- Isolated microcornea with normal ocular size
- Ocular colobomas without anophthalmia or microphthalmia

ICD-9-CM CODES (for case finding)

- ANOPHTHALMIA -- 743.00
- MICROPHTHALMIA -- 743.10-743.12

ICD-10-CM CODES (for case finding)

- OTHER ANOPHTHALMOS -- Q11.1
- MICROPHTHALMOS -- Q11.2

CBDRP CODES (for eligible defects)

- 743000-743004: Anophthalmos
- 743100-743104: Microphthalmos

4. Anotia/Microtia

BIRTH DEFECT & DEFINITION

- ANOTIA -- total absence of the external ear and canal
- MICROTIA -- malformation or hypoplasia of the auricle, ranging from measurably small external ear with minimal structural abnormality, to an ear with major structural alteration with absent or blind-ending canal

TYPES & DEFINITIONS

Microtia Classification System of Meurman (modified from Marks):

- TYPE I -- generally small ear that retains most of the overall structure of the normal auricle; similar to lop/ cup defect, auditory meatus is usually patent and defects of the ossicular chain are infrequent (Note: This is an exclusion – see below)
- TYPE II -- moderately severe anomaly with longitudinal mass of cartilage with some resemblance to pinna (rudimentary auricle will be hook-shaped, have an S-shape or question mark appearance)
- TYPE III -- ear is a rudiment of soft tissue and the auricle has no resemblance to the normal pinna
- TYPE IV -- complete absence of all external ear structures, anotia

NOTE: types I - III will occasionally be accompanied by preauricular tag(s)

INCLUSIONS

Standard (see section 1)

EXCLUSIONS

- Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect
- Small ears NOS or small ears that retain most of the normal structure
- Type I microtia with or without abnormality of the external auditory canal
- Isolated atresia or stenosis of the external auditory canal
- Normal ears that are misplaced: low set, posteriorly rotated, etc.
- “Decreased cartilage” reported as part of the estimate of gestational age

ICD-9-CM CODES (for case finding)

- ANOTIA -- 744.01
- OTHER ANOMALIES OF EXTERNAL EAR WITH IMPAIRMENT OF HEARING -- 744.02
- ABSENCE OF EAR, CONGENITAL -- 744.09
- MICROTIA -- 744.23

ICD-10-CM CODES (for case finding)

- CONGENITAL ABSENCE OF (EAR) AURICLE -- Q16.0

- CONGENITAL ABSENCE, ATRESIA AND STRICTURE OF AUDITORY CANAL (EXTERNAL) -- Q16.1
- CONGENITAL MALFORMATION OF EAR CAUSING IMPAIRMENT OF HEARING, UNSPECIFIED -- Q16.9
- MICROTIA -- Q17.2

CBD RP CODES (for eligible defects)

- 744010-744014: Anotia
- 744210-744214: Microtia

5-7. Conotruncal Heart Defects

Depending on the subtype, atresia of the pulmonary valve/artery is considered either among the conotruncal defects (8) or with the obstructive defects (10a, 10b).

BIRTH DEFECT & DEFINITION

- CONOTRUNCAL HEART DEFECTS -- anomalies of the outflow tract of the heart

TYPES & DEFINITIONS

5. TRUNCUS ARTERIOSUS (TA) -- single common arterial trunk instead of separate pulmonary artery and aorta, almost always associated with a malalignment-type VSD; there are subtypes 1, 2, 3 based on the pattern of truncal branching; no need to specify type
6. DEXTRO-TRANSPOSITION OF GREAT ARTERIES (DTGA, DTGV) -- transposed great arteries such that the pulmonary artery arises from the left ventricle and the aorta arises from the right ventricle
 - o May be isolated or with other congenital heart defects (e.g., VSD, pulmonic stenosis) -- If occurs with a VSD, do not code the VSD separately
7. TETRALOGY OF FALLOT (TOF, TET) -- tetralogy = a malalignment-type VSD creates subvalvar pulmonic stenosis, overriding of the aorta, and right ventricular hypertrophy (= 4 defects in one code)
 - o Includes TOF with "absent pulmonary valve"
 - o Includes PULMONARY ATRESIA WITH VSD (PA/VSD) -- atresia of the pulmonary valve/artery (under-developed connection from the right ventricle to the pulmonary artery) with membranous/malalignment-type VSD (NOT muscular or NOS type VSD); alternative archaic terms are Truncus, type 4, or pseudotruncus.
 - o "Pentalogy of Fallot" is an archaic term for TOF plus a secundum atrial septal defect (ASD2). If noted in the medical record, code both defects separately (TOF and ASD2).

INCLUSIONS

- Standard (see section 1)
- Cardiac defects must be confirmed by echocardiography (echo), catheterization (cath), surgery or autopsy; include cases diagnosed by prenatal ultrasound/echo only if done by a pediatric cardiologist or in a prenatal diagnosis center with expertise in this area.

EXCLUSIONS

- Cardiac defects that are clinically-diagnosed, i.e., using only physical exam, chest radiography, ECG are excluded
- Double Outlet Right Ventricle (DORV) of any type was an eligible defect in NBDPS, but it is NOT eligible for BD-STEPS. This includes all variants of DORV – those with

transposed/malposed great vessels and those with normally related great vessels but with anatomy similar to Tetralogy of Fallot (TOF).

- o In NBDPS, cases of DORV with anatomy similar to TOF were included in the Fallot (TOF) classification group. In BD-STEPS the Fallot category will not include these cases since DORV is ineligible. Thus, the Fallot category in NBDPS will NOT be exactly comparable to the Fallot category in BD-STEPS. To facilitate data combination between the two studies, NBDPS files will be created excluding the DORV-TOF cases from the Fallot category so the data can be combined and compared when desired.
- o In NBDPS, cases of DORV with transposed/malposed great arteries are already a separate category distinct from either DORV or Transposition of the Great Arteries (TGA). Thus, data combination of the TGA category between NBDPS and BDSTEPS will be unaffected by the ineligibility of DORV.
- PULMONARY ATRESIA WITH VSD (NOT TETRALOGY WITH PULMONARY ATRESIA) -- see Obstructive Heart Defects below
- PULMONARY ATRESIA WITH INTACT VENTRICULAR SEPTUM (PA/IVS) -- see Obstructive Heart Defects below

ICD-9-CM CODES (for case finding)

- TRUNCUS ARTERIOSUS -- 745.0
- DEXTRO-TRANSPOSITION OF GREAT ARTERIES -- 745.10
- OTHER TRANSPOSITION OF THE GREAT VESSELS -- 745.19
- TETRALOGY OF FALLOT -- 745.2
- PULMONARY ATRESIA WITH VSD
 - o Early versions of ICD-9-CM: (747.3 or 746.01) plus 745.4
 - o Later versions of ICD-9-CM: (747.31 or 746.01) plus 745.4

ICD-10-CM CODES (for case finding)

- COMMON ARTERIAL TRUNK -- Q20.0
- DISCORDANT VENTRICULOARTERIAL CONNECTION -- Q20.3
- DISCORDANT ATRIOVENTRICULAR CONNECTION -- Q20.5
- OTHER CONGENITAL MALFORMATIONS OF CARDIAC CHAMBERS AND CONNECTIONS -- Q20.8
- CONGENITAL MALFORMATIONS OF CARDIAC CHAMBERS AND CONNECTIONS, UNSPECIFIED -- Q20.9
- TETRALOGY OF FALLOT -- Q21.3
- PULMONARY VALVE ATRESIA WITH VSD -- Q22.0 plus Q21.0
- OTHER CONGENITAL MALFORMATIONS OF PULMONARY VALVE WITH VSD -- Q22.3 PLUS Q21.0
- ATRESIA OF PULMONARY ARTERY WITH VSD -- Q25.5 plus Q21.0

CBD RP CODES (for eligible defects)

- 745000: Truncus arteriosus (TA)

- 745100: Dextro-transposition of great arteries with intact ventricular septum (D-TGA/D-TGV w/ IVS) – Use this code if no VSD is present.
- 745110: Dextro-transposition of great arteries with ventricular septal defect (D-TGA/D-TGV w/ VSD) – Do not code the VSD separately.
- 745200: Tetralogy of Fallot (TOF)
 - o Do not code VSD and pulmonic stenosis separately
 - o 745200 + 746020 -- Tetralogy of Fallot (TOF) plus absent pulmonary valve
 - o 745200 + 745510 -- Pentalogy of Fallot (TOF plus ASD2)
- 747310: Pulmonary atresia with VSD (tetralogy of Fallot variant) (PA w/ VSD, TOF); Tetralogy of Fallot with pulmonary atresia

8-11. Obstructive Heart Defects

Depending on the subtype, atresia of the pulmonary valve/artery is considered either among the conotruncal defects (8) or with the obstructive defects (10a, 10b).

BIRTH DEFECT & DEFINITION

- OBSTRUCTIVE HEART DEFECTS--broad group of congenital heart defects in which there is obstruction to the flow of blood through either the left or right side of the heart or the great vessels

TYPES & DEFINITIONS

Right-Sided Obstructive Anomaly:

8. TRICUSPID ATRESIA (TriAtresia, TrA) -- atretic connection between the right atrium and the right ventricle, due to the absence or non-patency of the tricuspid valve
- 9a. PULMONARY ATRESIA WITH VSD (NOT TETRALOGY WITH PULMONARY ATRESIA) -- atresia of the pulmonary valve/artery (under-developed connection from the right ventricle to the pulmonary artery) with VSD described as "muscular" or "NOS" type (NOT described as "membranous/malalignment-type" types).
- 9b. PULMONARY ATRESIA WITH INTACT VENTRICULAR SEPTUM (PA/IVS) -- this is a distinctly different defect from pulmonary atresia with any type of VSD.

Left-Sided Obstructive Anomaly:

10. COARCTATION OF THE AORTA (COA) -- narrowing of the descending aorta, distal to the left subclavian artery; in most instances, the narrowing occurs close to the region where the ductus arteriosus inserts and is called juxtaductal coarctation
 - o Code separately, even in the presence of hypoplastic left heart syndrome
 - o There are no exclusions based on severity (even a 'mild' coarctation is included), although it is helpful to include information about the severity (gradient) or type (ledge vs. long segment coarctation) of the lesion
11. HYPOPLASTIC LEFT HEART SYNDROME (HLHS) -- extreme smallness of the left-sided heart structures (mitral valve and left ventricle) and aorta (including the aortic valve, ascending aorta, arch, and sometimes descending aorta [coarctation]); implies normally related great arteries
 - o Typical cases include mitral valve hypoplasia or atresia PLUS aortic hypoplasia or atresia, in the presence of a diminutive (non-apex forming) left ventricle
 - o In the typical case of HLHS, coarctation should be coded separately when present; mitral valve and aortic atresia or hypoplasia do not need separate coding if HLHS is coded
 - o A ventricular septal defect may be present and its size may influence the dimensions of the left ventricle (mitral atresia and intact septum are often associated with very small ventricle)

INCLUSIONS

- Standard (see section 1)
- Cardiac defects must be confirmed by echocardiography (echo), catheterization (cath), surgery or autopsy; except for coarctation of the aorta (see Exclusions below), include cases diagnosed by prenatal ultrasound/echo only if done by a pediatric cardiologist or in a prenatal diagnosis center with expertise in this area.

EXCLUSIONS

- Cardiac defects that are clinically-diagnosed, i.e., using only physical exam, chest radiography, ECG are excluded
- Tricuspid stenosis-- Review the case carefully to determine whether the abnormality is tricuspid atresia or tricuspid stenosis
- Interrupted aortic arch
- Coarctation of the aorta cases that are prenatally diagnosed but lack postnatal confirmation are excluded
- Unbalanced AV canal with right dominance in which the left ventricle and aorta may be small. Do not use the HLHS code.
- Pulmonary stenosis (valve or artery) with no mention of atresia.
- Ebstein's anomaly

ICD-9-CM CODES (for case finding)

- TRICUSPID ATRESIA (not stenosis) -- 746.1
- PULMONARY VALVE ATRESIA -- 746.01
- PULMONARY ARTERY ATRESIA WITH VSD
 - o Early versions of ICD-9-CM: 747.3 plus 745.4
 - o Later versions of ICD-9-CM: 747.31 plus 745.4
- COARCTATION OF THE AORTA -- 747.10
- HYPOPLASTIC LEFT HEART SYNDROME -- 746.7

ICD-10-CM CODES (for case finding)

- CONGENITAL TRICUSPID STENOSIS -- Q22.4
 - o Review the case carefully to determine whether the abnormality is tricuspid atresia or tricuspid stenosis
- PULMONARY VALVE ATRESIA -- Q22.0
- OTHER CONGENITAL MALFORMATIONS OF PULMONARY VALVE WITH VSD -- Q22.3 PLUS Q21.0
- ATRESIA OF PULMONARY ARTERY WITH VSD -- Q25.5 plus Q21.0
- COARCTATION OF AORTA -- Q25.1
- ATRESIA OF AORTA -- Q25.2
- HYPOPLASTIC RIGHT HEART SYNDROME -- Q22.6
- HYPOPLASTIC LEFT HEART SYNDROME -- Q23.4

CBDRP CODES (for eligible defects)

- 746100: Tricuspid atresia (TrA)
- 746000: Pulmonary valve atresia/intact ventricular septum (PA/IVS)

- 746030: Pulmonary valve atresia with VSD (not tetralogy of Fallot variant) (PA w/ VSD, not TOF)
- 747100: Coarctation of the aorta, preductal (proximal)
- 747110: Coarctation of the aorta, postductal (distal)
- 747120: Coarctation of the aorta, juxtaductal
- 747190: Coarctation of the aorta, NOS
- 746700: Hypoplastic left heart syndrome (HLHS).
-

12. Total Anomalous Pulmonary Venous Connection

BIRTH DEFECT & DEFINITION

- TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION --a condition in which all four of the pulmonary veins fail to connect to the left atrium. They drain anomalously into the right side of the heart or into other vessels instead of into the left side of the heart; often occurs with other cardiac defects. Pulmonary blood can return to the heart via supra-diaphragmatic or infra-diaphragmatic routes; these details are not needed for coding purposes.

TYPES & DEFINITIONS

These are all synonyms:

- TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION (TAPVC)
- TOTAL ANOMALOUS PULMONARY VENOUS RETURN (TAPVR)
- TOTAL ANOMALOUS PULMONARY DRAINAGE (TAPVD)

INCLUSIONS

- Standard (see section 1)
- Cardiac defects must be confirmed by echocardiography (echo), catheterization (cath), surgery or autopsy; include cases diagnosed by prenatal ultrasound/echo only if done by a pediatric cardiologist or in a prenatal diagnosis center with expertise in this area.

EXCLUSIONS

- Cardiac defects that are clinically-diagnosed, i.e., using only physical exam, chest radiography, ECG are excluded
- Cases prenatally diagnosed that do not have a postnatal diagnostic examination to confirm the diagnosis
- PARTIAL ANOMALOUS PULMONARY VENOUS CONNECTION/RETURN/DRAINAGE -- a condition in which 1, 2, or 3 pulmonary veins fail to connect to the left atrium but others do connect.

ICD-9-CM CODES (for case finding)

- TOTAL ANOMALOUS PULMONARY VENOUS RETURN -- 747.41

ICD-10-CM CODES (for case finding)

- TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION -- Q26.2
- ANOMALOUS PULMONARY VENOUS CONNECTION, UNSPECIFIED -- Q26.4

CBDRP CODES (for eligible defects)

- 747420: Total anomalous pulmonary venous return/connection/drainage (TAPVR)

13. Cleft Lip +/- Palate

BIRTH DEFECT & DEFINITION

- CLEFT LIP +/- PALATE -- incomplete closure of the lip; often accompanied by a maxillary alveolar (gum) defect and/or cleft palate; maxillary alveolar defect may be a complete cleft that is continuous with the cleft palate, or it may be limited to a notch on the gum; cleft lip may be unilateral, bilateral, or median; median cleft lip is distinguished from bilateral cleft lip by agenesis of the premaxilla.

TYPES & DEFINITIONS

- COMPLETE CLEFT LIP -- defect that extends through the entirety of the lip and the nasal floor; may be unilateral or bilateral; usually associated with a more severe nasal deformation.
- INCOMPLETE CLEFT LIP -- defect of the lip that does not extend into the nasal floor; may be unilateral or bilateral; there may be an incomplete cleft lip on one side and a complete cleft lip on the other side.
- PSEUDOCLEFT LIP -- an abnormal linear thickening, depressed groove, or subtle scar-like pigmentary difference of the skin paralleling the philtral ridge on the affected side; may be associated with a slight notch of the vermilion or a mild slouching of the alar cartilage.

INCLUSIONS

- Standard (see section 1)
- If cleft palate is associated with any type of cleft lip, it is coded as a cleft lip and palate, not cleft palate

EXCLUSIONS

- Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect
- Pseudocleft lip; microform cleft lip; forme fruste cleft lip
- Tessier type facial clefts
- Oblique facial clefts
- Cleft gum only
- Any cleft lip with coexisting holoprosencephaly (include under holoprosencephaly only)
- Any cleft lip with coexisting anencephaly

ICD-9-CM CODES (for case finding)

- CLEFT LIP -- 749.10-749.14
- CLEFT LIP WITH PALATE -- 749.20-749.25

ICD-10-CM CODES (for case finding)

- CLEFT LIP -- Q36.0-Q36.9
- CLEFT PALATE WITH CLEFT LIP -- Q37.0-Q37.9

CBD RP CODES (for eligible defects)

- 749101-749103: Cleft lip, unilateral
- 749110: Cleft lip, bilateral
- 749120: Cleft lip, central
- 749195: Cleft lip, NOS
- 749201-749203: Cleft lip and palate, unilateral
- 749210: Cleft lip and palate, bilateral cleft lip
- 749220: Cleft lip and palate, central cleft lip
- 749290: Cleft lip and palate, NOS

14. Cleft Palate

BIRTH DEFECT & DEFINITION

- CLEFT PALATE -- a hole or opening in the roof of the mouth due to incomplete fusion of the palatal shelves; may be limited to the soft palate only or also extend into hard palate

TYPES & DEFINITIONS

- PIERRE ROBIN ANOMALY (SEQUENCE) -- combination of micrognathia, cleft palate, glossoptosis (tongue falls back into the pharynx)
- SUBMUCOUS CLEFT PALATE -- a defect of the soft palate with the mucosa or a reduced, thin muscle layer bridging across it; difficult to diagnose clinically in the 1st year; often associated with a bifid uvula

INCLUSIONS

- Standard (see section 1)
- PIERRE ROBIN ANOMALY (SEQUENCE)

EXCLUSIONS

- Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect
- Submucous cleft palate
- Bifid or cleft uvula without overt cleft palate
- Any cleft palate with coexisting holoprosencephaly (include under holoprosencephaly only)
- Any cleft palate with coexisting anencephaly
- If cleft palate is associated with any cleft lip, it is coded as a cleft lip and palate, not cleft palate.

ICD-9-CM CODES (for case finding)

- CLEFT PALATE -- 749.00-749.04

ICD-10-CM CODES (for case finding)

- CLEFT PALATE -- Q35.1-Q35.9, excluding CLEFT UVULA Q35.7

CBD RP CODES (for eligible defects)

- 749001-749003: Cleft hard palate, unilateral
- 749010: Cleft hard palate, bilateral
- 749020: Cleft hard palate, central
- 749030: Cleft hard palate, NOS
- 749041-749043: Cleft soft palate, unilateral
- 749050: Cleft soft palate, bilateral
- 749060: Cleft soft palate, central
- 749070: Cleft soft palate, NOS
- 749090: Cleft palate, NOS

15. Esophageal Atresia +/- TE Fistula

BIRTH DEFECT & DEFINITION

- ESOPHAGEAL ATRESIA +/- TRACHEOESOPHAGEAL FISTULA (T-E FISTULA, TEF) -- congenital complete discontinuity of the lumen of the esophagus resulting in a blind esophageal pouch occurring with or without an abnormal communication between the esophagus and the trachea

TYPES & DEFINITIONS

- There are several classification schemas.
- In 90% of cases the upper esophagus ends in a blind pouch and the lower segment forms a fistula with the trachea.

INCLUSIONS

Standard (see section 1)

EXCLUSIONS

- Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect
- Tracheoesophageal fistula without esophageal atresia
- Esophageal stenosis
- Trachea atresia
- Tracheoesophageal cleft

ICD-9-CM CODES (for case finding)

- ESOPHAGEAL ATRESIA, TRACHEOESOPHAGEAL FISTULA -- 750.3

ICD-10-CM CODES (for case finding)

- ATRESIA OF ESOPHAGUS WITHOUT FISTULA -- Q39.0
- ATRESIA OF ESOPHAGUS WITH TRACHEO-ESOPHAGEAL FISTULA -- Q39.1
- CONGENITAL TRACHEO-ESOPHAGEAL FISTULA WITHOUT ATRESIA -- Q39.2

CBDRP CODES (for eligible defects)

- 750300: Esophageal atresia without TE fistula
- 750310: Esophageal atresia with TE fistula

16. Limb Deficiency, Transverse

BIRTH DEFECT & DEFINITION

- TRANSVERSE LIMB DEFICIENCY--Complete or partial absence of the distal structures of a limb in a transverse plane at the point where the deficiency begins.

TYPES & DEFINITIONS

- AMELIA -- complete absence of a limb
- TRANSVERSE LIMB REDUCTION DEFECT -- a synonym for transverse limb deficiency
- CONGENITAL AMPUTATION -- a synonym for transverse limb deficiency
- HEMI- OR MEROMELIA -- partial absence of a limb (a rather nonspecific; term can also be used for longitudinal defects)
- TRANSVERSE TERMINAL DEFICIENCY -- absence of distal structures with proximal structures essentially intact (used for deficiencies below the elbow)
- APHALANGIA -- absence of phalanges
- ADACTYLY -- absence of digits
- OLIGODACTYLY -- fewer than 5 digits
- ACHEIRIA -- absence of a hand
- AMNION RUPTURE SEQUENCE -- limb deficiencies and constrictions associated with tears or rupture in the amnion

INCLUSIONS

- Standard (see section 1)
- Transverse limb deficiencies that are secondary to amnion rupture sequence or amniotic bands
- Transverse limb deficiencies with rudimentary or “nubbin” fingers attached to the stump. NOTE: When “nubbins” are present include a description of their location, number and size with the description of the limb deficiency

EXCLUSIONS

- Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect
- Isolated missing digits where the rest of the hand and proximal limb are intact
- Unspecified type of limb deficiency
- Generalized limb shortening including chondrodysplasias
- Nail hypoplasia
- Brachydactylies type A-E
- Lower extremity deficiencies with sirenomelia sequence
- Absent digits with split-hand or split-foot
- Absent digits with longitudinal deficiencies (absent digits with corresponding metacarpal/metatarsal +/- long bone deficiencies, e.g., radial, ulnar, tibial, or fibular ray defects)
- Isolated absent (missing) or hypoplastic thumb or great toe not associated with other defects of the same limb

- LIMB-BODY WALL COMPLEX -- disruption complex involving body wall defects and limb deficiencies, as well as neural tube defects, heart, and other anomalies
- Phocomelia defects
- Intercalary defects
- Longitudinal limb deficiencies
- Amnion rupture sequence or amniotic bands not associated with a transverse limb deficiency

ICD-9-CM CODES (for case finding)

- UPPER LIMB TRANSVERSE DEFICIENCY -- 755.21
- UPPER LIMB LONGITUDINAL DEFICIENCY OF PHALANGE(S) -- 755.29
- LOWER LIMB TRANSVERSE DEFICIENCY -- 755.31
- LOWER LIMB LONGITUDINAL DEFICIENCY OF PHALANGE(S) -- 755.39

ICD-10-CM CODES (for case finding)

- REDUCTION DEFECTS OF UPPER LIMB -- Q71.00-Q71.899,
- REDUCTION DEFECTS OF LOWER LIMB -- Q72.00-Q72.899
- REDUCTION DEFECTS OF UNSPECIFIED LIMB -- Q73.0-Q73.8

CBDRP CODES (for eligible defects)

- 755200-755204: Transverse deficiency or amputation of the arm, NOS
- 755205-755209: Total absence of the arm
- 755240-755244: Absence of the forearm and hand
-
- 755285: Absence of hand, Laterality unknown
- 755286: Absence of hand, Left
- 755287: Absence of hand, Right
- 755288: Absence of hand, Unilateral, side unknown
- 755289: Absence of hand, Bilateral
- 755300-755304: Transverse deficiency or amputation of the leg, NOS
- 755305-755309: Total absence of the lower limb
- 755340-755344: Absence of the lower leg and foot
-
- 755385: Absence of foot, Laterality unknown
- 755386: Absence of foot, Left
- 755387: Absence of foot, Right
- 755388: Absence of foot, Unilateral, side unknown
- 755389: Absence of foot, Bilateral

17. Diaphragmatic Hernia

BIRTH DEFECT & DEFINITION

- DIAPHRAGMATIC DEFECTS (HERNIA) -- incomplete formation of the diaphragm through which some portion of the abdominal contents herniates into the thoracic cavity

TYPES & DEFINITIONS

- CONGENITAL DIAPHRAGMATIC HERNIA -- This is a synonym for a diaphragmatic defect.
- POSTEROLATERAL HERNIA, also known as a BOCHDALEK HERNIA -- defect involving the posterior and/or lateral portions of the diaphragm
- AGENESIS, ABSENCE, or APLASIA OF THE DIAPHRAGM -- apparent absence of an entire side of the diaphragm; represents a large Bochdalek hernia
- ANTERIOR HERNIA, also known as a MORGAGNI HERNIA, RETROSTERNAL HERNIA, PARASTERNAL HERNIA, MORGAGNI-LARREY HERNIA -- defect involving the anterior portion of the diaphragm
- LARGE ANTERIOR HERNIA, also known as a SEPTUM TRANSVERSUM HERNIA -- type of diaphragmatic defect found in Pentalogy of Cantrell
- PARAESOPHAGEAL HERNIA -- defect in the diaphragm surrounding the esophagus
- HEMIDIAPHRAGM -- This is a synonym for a diaphragmatic defect.
- OTHER -- includes, for example, central diaphragm defects, anterolateral defects, and unusual/atypical defects
- HERNIA SAC -- approximately 15% of CDH have a sac, which is a localized thinning or out-pouching of the diaphragm; a sac is not a type of hernia.

INCLUSIONS

- Standard (see section 1)
- Prenatally diagnosed cases should be included only if bowel was documented in the chest by prenatal ultrasound
- Diaphragmatic hernia with Pentalogy of Cantrell

EXCLUSIONS

- Eventration of the diaphragm -- not a true herniation, but an upward displacement of abdominal contents into an out-pouched diaphragm resulting from weakness or absence of diaphragmatic musculature
- Hiatal hernia
- CCAM (cystic adenomatoid malformation of the lung)
- Hernia sac not associated with an actual defect in the diaphragm

ICD-9-CM CODES (for case finding)

- DIAPHRAGMATIC HERNIA -- 756.6

ICD-10-CM CODES (for case finding)

- CONGENITAL DIAPHRAGMATIC HERNIA -- Q79.0

- ABSENCE OF DIAPHRAGM -- Q79.1

CBD RP CODES (for eligible defects)

- 756600-756604: Diaphragmatic hernia, NOS
- 756605: Diaphragmatic hernia, esophageal
- 756610-756614: Diaphragmatic hernia, Bochdalek
- 756615-756619: Diaphragmatic hernia, Morgagni

18. Gastroschisis

BIRTH DEFECT & DEFINITION

- GASTROSCHISIS--congenital fissure of the anterior abdominal wall, lateral to the umbilicus usually to the right, with a small bridge of skin separating the defect from the umbilicus; accompanied by herniation of the small, and part of the large, intestines, and occasionally other abdominal organ, into the amniotic cavity, and lacking a protective membrane

TYPES & DEFINITIONS

- LIMB-BODY WALL COMPLEX -- disruption complex involving body wall defects and limb deficiencies, as well as neural tube defects, heart, and other anomalies (Note: This is an exclusion - see below)

INCLUSIONS

- Standard (see section 1)
- Prenatally diagnosed cases if high resolution ultrasound was done and the umbilicus was visualized to be separate from the defect

EXCLUSIONS

- Gastroschisis associated with LIMB-BODY WALL COMPLEX

ICD-9-CM CODES (for case finding)

- GASTROSCHISIS -- 756.73

ICD-10-CM CODES (for case finding)

- GASTROSCHISIS -- Q79.3

CBDRP CODES (for eligible defects)

- 756710: Gastroschisis

19. Microcephaly with a Coexisting Brain Anomaly

BIRTH DEFECT & DEFINITION

MICROCEPHALY – the clinical finding of a small head circumference (HC) when compared with infants of the same sex and age. It is the intent of BD-STEPS II to ascertain phenotypes including microcephaly that may be associated with congenital infections. Therefore, for the purposes of BD-STEPS II, the diagnosis of microcephaly must be associated with at least one other major congenital brain abnormality.

TYPES & DEFINITIONS

Mention of microcephaly or a small head must be noted in the medical record AS WELL AS diagnosis of at least one other major congenital brain anomaly. The diagnosis of microcephaly should not be assigned by surveillance staff based on the HC measurement alone without mention of microcephaly or a small head in the medical record.

For the purposes of BD-STEPS II, microcephaly is defined as:

- For live births, the measured HC adjusted for gestational age and sex must be less than 3rd percentile at birth or, if not measured at birth, within the first 2 weeks of life.
- For pregnancy losses, the HC measured on a prenatal ultrasound must be more than 3 standard deviations below the mean; OR, the measured postnatal HC adjusted for gestational age and sex must be less than 3rd percentile at delivery.
- BD-STEPS II adopts the CDC growth charts as the standard when determining HC percentiles. Separate charts for boys and girls ages birth to 36 months are available at: https://www.cdc.gov/growthcharts/clinical_charts.htm

NOTE: Please identify all HC values recorded within the first 2 weeks of life. If multiple HCs are recorded, all must measure less than the 3rd percentile for sex and gestational age at birth to be included.

INCLUSIONS

Microcephaly AND at least one of the following brain abnormalities diagnosed by fetal MRI, postnatal imaging (cranial ultrasound, CT, MRI), or autopsy:

- Intracranial calcifications – calcifications noted anywhere within the substance of the brain, often described as brightly echogenic foci on ultrasound, CT, or MRI scan
- Cerebral or cortical atrophy – atrophy of any part of the cerebral hemispheres, including cerebral atrophy, cortical atrophy, or cortical loss
- Abnormal cortical gyral patterns diagnosed after 28 weeks gestation – includes lissencephaly, agyria, smooth brain, pachygyria, macrogyria, incomplete lissencephaly (reduced number of gyri which are wider than normal), polymicrogyria, gray matter heterotopia, ectopia or marginal glioneuronal heterotopias, leptomenigeal heterotopias, neuronal migration disorder, neuronal maturation disorder, schizencephaly
- Corpus callosum abnormalities – includes agenesis or absence (either complete or partial), hypoplasia, dysgenesis, or thinning

- Cerebellar abnormalities – includes agenesis, hypoplasia, dysplasia, atrophy, Dandy Walker malformation, Dandy Walker Blake continuum, Dandy Walker variant
- Porencephaly – includes porencephalic cyst or cavity, developmental porencephaly
- Hydranencephaly – includes unilateral or bilateral
- Moderate or severe ventriculomegaly or hydrocephaly, including communicating hydrocephaly and that due to aqueductal stenosis
- Fetal brain disruption sequence – components are severe microcephaly, overlapping cranial sutures, prominent occipital bone, redundant scalp skin or rugae, in addition to severe neurologic impairment. All components must be present to make the diagnosis.
- Septo-optic dysplasia – coexisting hypoplasia of the optic nerve and/or optic chiasma, absence of the septum pellucidum, abnormalities of the corpus callosum
- Multiple or bilateral ependymal or subependymal cysts

NOTE: While microcephaly must be diagnosed within the first 2 weeks of life to qualify, an associated brain abnormality can be diagnosed at any time.

EXCLUSIONS

- Isolated microcephaly without a coexisting major brain abnormality
- Mention of microcephaly in the medical record with no HC measurement documented or with a HC measurement >3% for gestational age and sex at birth or within the first 2 weeks of life
- Microcephaly first mentioned beyond two weeks of age
- Infants with more than one HC recorded within the first 2 weeks of life for which at least one HC measures more than the 3rd percentile for gestational age and sex at birth.
- Acquired microcephaly – examples include microcephaly resulting from a delivery complication, postnatal insult or trauma, meningitis, etc.
- Macrocephaly, megalencephaly, macroencephaly
- Microcephaly associated with one or more of the following only:
 - Neural tube defect (spina bifida, anencephaly, encephalocele, and associated abnormalities). Neural tube defects are not known to be associated with congenital infections.
 - Holoprosencephaly
 - Brain tumor
 - Intracranial calcification resulting from a brain tumor or thrombosis in a large cerebral blood vessel, such as might be seen with tuberous sclerosis or a transverse/straight sinus thrombosis
 - Cerebral or cortical atrophy that is secondary to prematurity, a delivery complication, or postnatal insult
 - Abnormal cortical gyral patterns diagnosed before 28 weeks gestation and not confirmed after 28 weeks
 - Arnold-Chiari malformation, including Chiari malformation type II
 - Mega cisterna magna
 - Prenatal or postnatal intraventricular hemorrhage (IVH) and resulting abnormalities

- o Periventricular leukomalacia
- o Encephalomalacia
- o Porencephaly resulting from prematurity or a delivery complication – examples include a severe hypoxic/ischemic event at delivery, postnatal IVH, periventricular leukomalacia, encephalomalacia, etc.
- o Arachnoid or choroid plexus cyst
- o Ventriculomegaly or hydrocephalus that is secondary to another qualifying brain abnormality such as spina bifida or holoprosencephaly
- o Ventriculomegaly or hydrocephalus that is secondary to a non-qualifying brain abnormality such as obstruction from an arachnoid cyst
- o Ventriculomegaly or hydrocephalus described as mild, borderline, or prominent ventricles without further qualifier
- o Slit-like ventricles or asymmetrical ventricles without mention of ventriculomegaly or hydrocephalus
- o Single isolated ependymal or subependymal cyst
- o Abnormalities of the vasculature of the brain
- o Cranial nerve abnormalities without another qualifying brain abnormality
- o Isolated absence of the septum pellucidum without another qualifying brain abnormality
- Microcephaly associated with a brain abnormality that is diagnosed solely by prenatal ultrasound without confirmation by postnatal imaging or autopsy exam

ICD-9-CM CODES (for case finding)

- MICROCEPHALUS -- 742.1
- MAJOR CONGENITAL BRAIN ANOMALIES -- 742.2-742.4

ICD-10-CM CODES (for case finding)

- MICROCEPHALY -- Q02
- MAJOR CONGENITAL BRAIN ANOMALIES -- Q03.0- Q04.9

CBD RP CODES (for eligible defects)

- 742100: Microcephalus
- 742200: Fetal brain disruption sequence
- 742210: Anomalies of corpus callosum
- 742230: Anomalies of cerebellum, excluding Dandy-Walker malformation/variants (use code 742310)
- 742235: Hypoplasia of other parts of the cerebellum with or without hypoplasia of the vermis
- 742236: Hypoplasia of the cerebellar vermis only (**NOTE: THIS IS A NEW CODE UNIQUE TO BD-STEPS II**)
- 742240: Agyria, smooth brain, lissencephaly
- 742245: Other abnormal gyral patterns, including pachygyria, incomplete lissencephaly, macrogyria (**NOTE: THIS IS A NEW CODE UNIQUE TO BD-STEPS II**)
- 742250: Microgyria, polymicrogyria

- 742255: Schizencephaly (**NOTE: THIS IS A NEW CODE UNIQUE TO BD-STEPS II**)
- 742256: Other neuronal migration/maturation disorders, heterotopia (**NOTE: THIS IS A NEW CODE UNIQUE TO BD-STEPS II**)
- 742280: Septo-optic dysplasia
- 742300: Hydrocephalus due to anomalies of aqueduct of Sylvius
- 742310: Dandy-Walker malformation/variants
- 742320: Hydranencephaly
- 742380: Hydrocephalus, other specified
- 742390: Hydrocephalus, NOS
- 742410: Porencephaly, Laterality unknown
- 742411: Porencephaly, Left side of brain only
- 742412: Porencephaly, Right side of brain only
- 742413: Porencephaly, Unilateral, side unknown
- 742414: Porencephaly, Both sides of brain
- 742420: Ependymal or subependymal cysts, Laterality unknown
- 742421: Ependymal or subependymal cysts, Left side of brain
- 742422: Ependymal or subependymal cysts, Right side of brain
- 742423: Ependymal or subependymal cysts, Unilateral, side unknown
- 742424: Ependymal or subependymal cysts, Both sides of brain
- 742430: Intracranial calcification (**NOTE: THIS IS A NEW CODE UNIQUE TO BD-STEPS II**)
- 742480: Cerebral/cortical atrophy and/or hydrocephalus *ex vacuo*
- 742.486: Small brain

20. Holoprosencephaly

BIRTH DEFECT & DEFINITION

HOLOPROSENCEPHALY—structural brain anomaly that results from variable degrees of incomplete cleavage of the prosencephalon (embryonic forebrain), which fails to cleave sagittally into the right and left cerebral hemispheres and transversely into telencephalon and diencephalon.

TYPES & DEFINITIONS

Anatomic subgroups:

- Alobar holoprosencephaly – complete lack of division of the cerebral hemispheres, resulting in one single ventricle instead of right and left lateral cerebral ventricles
- Semi-lobar holoprosencephaly – partial division of the cerebral hemispheres, with absence of olfactory bulbs, absence of corpus callosum, and underdeveloped (rudimentary) lobes of the brain
- Lobar holoprosencephaly – cerebral hemispheres are mostly divided but remain fused in the front
- Middle interhemispheric variant of holoprosencephaly (MIHV) – lack of division of the posterior frontal and parietal lobes of the brain
- Arhinencephaly – an older term for holoprosencephaly which refers more specifically to structural defects of the olfactory system or nose
- Holotelencephaly – holoprosencephaly with associated arhinencephaly

Associated facial phenotypes:

- Cyclopia – a form of holoprosencephaly where a single, central eye is present
- Cebocephaly – a form of holoprosencephaly where the nose is underdeveloped (e.g., single nostril; proboscis) and closely set eyes (hypotelorism) are present
- Ethmocephaly – a form of holoprosencephaly where the eyes are closely set (hypotelorism), the nose is usually absent, and a proboscis is present
- Premaxillary agenesis (medial cleft lip) – midline cleft lip with absent premaxilla; nose small and flat, and ocular hypotelorism common
- Less severe facial features – hypotelorism or hypertelorism, flat nose, unilateral or bilateral cleft lip

INCLUSIONS

Standard (see section 1)

EXCLUSIONS

- Aprosencephaly
- Atelencephaly
- Hydranencephaly
- Arhinencephaly without holoprosencephaly

ICD-9-CM CODES (for case finding)

- REDUCTION DEFORMITIES OF BRAIN—742.2

ICD-10-CM CODES (for case finding)

- HOLOPROSENCEPHALY—Q04.2
- ARHINENCEPHALY—Q04.1

CBD RP CODES (for eligible defects)

- 742260: Holoprosencephaly, NOS
- 742265: Holoprosencephaly, alobar
- 742266: Holoprosencephaly, semilobar
- 742267: Holoprosencephaly, lobar

21. Anterior Segment Eye Defects

BIRTH DEFECT & DEFINITION

ANTERIOR SEGMENT EYE DEFECTS – defects of the anterior chamber of the eye including the lens. Any history of eye defects, particularly for parents or siblings with cataracts, should be abstracted.

TYPES & DEFINITIONS

- CATARACT--a loss of transparency of the crystalline lens of the eye or of its capsule; includes opacities affecting any part of the lens, including anterior, posterior and zonular cataracts
- APHAKIA--absence of the lens
- LENS COLOBOMA--absence of a portion of the lens
- MICROSPHEROPHAKIA--a small, spherically-shaped lens; can be seen as microphakia (small lens) or spherophakia (spherically-shaped lens)
- CONGENITAL GLAUCOMA OR BUPHTHALMOS--disease of the eye characterized by increased intraocular pressure, often due to restricted outflow of the aqueous humor through Schlemm's canal
- ANIRIDIA--absence of the iris
- INTRAOCULAR CALCIFICATIONS – Calcifications within the anterior segment of the eye

INCLUSIONS

- All cases must include diagnosis by an ophthalmologist within 12 months of life or confirmation by surgical pathology or autopsy.
- Cases with isolated cataracts in which the family history appears consistent with a Mendelian inheritance pattern are included, but may be excluded for certain analyses.

EXCLUSIONS

- Cataracts that occur as a result of ophthalmologic surgery or trauma
- Minor lens opacities that are not clinically significant (i.e., requiring no intervention or follow-up)
- Cases that are not diagnosed by an ophthalmologist, surgical pathology or autopsy, or for whom diagnosis was made after age 1 year
- Opacities of the cornea (clear transparent membrane covering the front of the eye over the iris)
- Glaucoma co-occurring with anencephaly, holoprosencephaly, anterior encephaloceles, or amniotic bands
- Isolated microcornea with normal ocular size
- Iris coloboma without any other eligible anterior chamber defect
- Cases with anterior segment dysgenesis, Rieger anomaly, Peters anomaly, Peters Plus syndrome, Axenfeld anomaly, Alagille syndrome, Velocardiofacial/DiGeorge syndrome, or Sturge-Weber syndrome

ICD-9-CM CODES (for case finding)

- CATARACT AND LENS ANOMALIES -- 743.3
- GLAUCOMA -- 743.2
- ANTERIOR SEGMENT ANOMALIES -- 743.44-743.46

ICD-10-CM CODES (for case finding)

- CATARACT AND LENS ANOMALIES - Q12.0-Q12.9
- GLAUCOMA -- Q15.0
- ANTERIOR SEGMENT ANOMALIES -- Q13.1-Q13.2, Q13.8-Q13.9

CBD RP CODES (for eligible defects)

- 743200-743204: Buphthalmos, congenital glaucoma
- 743300-743304: Absence of lens, congenital aphakia
- 743310-743314: Spherical lens, spherophakia
- 743320-743324: Cataract, NOS
- 743340-743344: Coloboma of lens
- 743350-743354: Cataract, anterior polar
- 743360-743364: Cataract, other specified
- 743420-743424: Absence of iris, aniridia

(NOTE: THESE ARE NEW CODES UNIQUE TO BD-STEPS II):

- 743475: Intraocular calcification of the anterior segment, Laterality unknown
- 743476: Intraocular calcification of the anterior segment, Left eye only
- 743477: Intraocular calcification of the anterior segment, Right eye only
- 743478: Intraocular calcification of the anterior segment, Unilateral, side unknown
- 743479: Intraocular calcification of the anterior segment, Both eyes

22. Posterior Segment Eye Defects

BIRTH DEFECT & DEFINITION

POSTERIOR SEGMENT EYE DEFECTS – defects of the posterior chamber of the eye including the retina, choroid, optic disc and optic nerve.

TYPES & DEFINITIONS

- Optic nerve hypoplasia
- Chorioretinal atrophy, scarring, or pigmentary anomaly
- Colobomas of the retina, choroid, optic nerve or optic disc
- Intraocular calcification

INCLUSIONS

- All cases must include diagnosis by an ophthalmologist within 12 months of life or confirmation by surgical pathology or autopsy.

EXCLUSIONS

- Immature retina
- Persistent fetal vasculature
- “Bear track” pigmentary changes of the retina
- Retinopathy of prematurity
- Retinal hemorrhage due to trauma, including delivery
- Optic nerve pallor alone without evidence/mention of hypoplasia
- Retinal pigment epithelial hypertrophy

ICD-9-CM CODES (for case finding)

- POSTERIOR SEGMENT ANOMALIES -- 743.51-743.57

ICD-10-CM CODES (for case finding)

- POSTERIOR SEGMENT ANOMALIES -- Q14.0-Q14.9

CBD RP CODES (for eligible defects)

- 743500: Intraocular calcification of the posterior segment
- 743510-743514: Chorioretinal atrophy, scarring, or pigmentary anomaly
- 743520-743524: Optic nerve hypoplasia
- 743580-743584: Coloboma of the retina, choroid, optic nerve or optic disc

22. Arthrogryposis

BIRTH DEFECT & DEFINITION

ARTHROGRYPOSIS – congenital contracture of major joints in two or more different areas of the body. Most joints are flexed, but some can be extended. The contractures can be fixed or more flexible. Muscles of the affected limbs may be atrophied or underdeveloped.

TYPES & DEFINITIONS

INCLUSIONS

- Must involve major joints in two or more different areas of the body
- Distal arthrogryposis—involves just the hands and feet
- Arthrogryposis multiplex congenital (AMC)
- Multiple pterygia – the contractures are accompanied by webbing of the skin across the affected joints

EXCLUSIONS

- Non-fixed, reducible positioning of the limbs or joints that can easily be moved to their typical neutral position
- Isolated finger contractures without contractures of other joints
- Isolated clubfoot, unilateral or bilateral
- Posturing of the limbs in the flexed position due to increased muscle or nerve tone (hypertonia)
- Contractures associated with musculoskeletal limb abnormalities
- Isolated clenched hand or fist, unilateral or bilateral
- Contracture of a single major joint only or of multiple major joints in the same area of the body (e.g., contracture of a wrist and elbow only)

ICD-9-CM CODES (for case finding)

- ARTHROGRYPOSIS MULTIPLEX CONGENITA -- 754.89

ICD-10-CM CODES (for case finding)

- ARTHROGRYPOSIS MULTIPLEX CONGENITA -- Q74.3

CBD RP CODES (for eligible defects)

- 755800: Arthrogryposis multiplex congenita

Heterotaxy (Exclusion)

If a case has an eligible heart defect but also meets the criteria for heterotaxy as defined below by Lin, et al., Am J Med Genet Part A. 2014;164A:2581–2591, the case is excluded from BD-STEPS II. If a case has an eligible heart defect but does not meet the criteria below, the case is included in BD-STEPS regardless of whether there is mention of heterotaxy, isomerism, situs abnormality, or laterality defect in the medical record.

Classification of heterotaxy requires at least 3 of the following features selected from groups 1-8:

1. Characteristic congenital heart defects
 - Pulmonary venous anomalies
 - Totally anomalous pulmonary venous connection or drainage
 - Partially anomalous pulmonary venous connection or drainage
 - Atrial anomalies
 - Atrial situs ambiguous or situs inversus
 - Common atrium
 - Common atrioventricular canal or atrioventricular septal defects
 - Complete atrioventricular canal defect
 - Partial atrioventricular canal defect
 - Transitional atrioventricular canal defect
 - Ventricular abnormalities
 - Hypoplastic or single left ventricle
 - Hypoplastic or single right ventricle
 - Ventricular malposition (e.g., L-loop, superior-inferior, criss-cross)
 - Ventriculo arterial alignment abnormalities
 - Double-outlet ventricle
 - D-loop transposition of great arteries
 - L-loop transposition of great arteries
 - Truncus arteriosus
 - Tetralogy of Fallot (including TOF/PS, TOF/PA, and TOF/APV)
 - Ventricular outflow abnormalities
 - Subvalvular or valvar pulmonary stenosis
 - Pulmonary atresia with intact ventricular septum
 - Pulmonary atresia with ventricular septal defect (not TOF-type)
 - Subvalvar or valvar aortic stenosis
 - Coarctation of the aorta
2. Biliary atresia
3. Abdominal situs abnormality
 - Abdominal situs inversus
 - Situs ambiguous (midline or transverse liver, midline aorta, ipsilateral aorta and inferior vena cava)

4. Spleen abnormality (confirmed by imaging, autopsy, or by Howell-Jolly bodies)
 - Asplenia
 - Polysplenia
 - Single right-sided spleen

5. Isomerism of bronchi
 - Bilateral left bronchial morphology
 - Bilateral right bronchial morphology

6. Isomerism of the lungs
 - Bilateral two lobes (left-sidedness)
 - Bilateral three lobes (right-sidedness)

7. Similar morphology of the atria appendages (“isomerism”)

8. Two of the following
 - Systemic venous anomalies
 - Bilateral superior vena cava
 - Interrupted inferior vena cava
 - Unroofed (absent) coronary sinus
 - Intestinal malrotation
 - Malrotation, nonrotation of the colon
 - Malrotation of the small intestine
 - Absent gallbladder